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FACT SHEET

tuberous sclerosis

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What is tuberous sclerosis?

Tuberous sclerosis (TS) is a genetic disorder that causes a variety of neurological and physical symptoms, including seizures, mental retardation, and tumors of the brain, kidneys, eyes, or other organs.

Other signs of the disorder include

- White spots on the skin of the trunk and limbs
- Characteristic bumpy skin rash over the cheeks below the eyes
- Delayed development of speech and language skills
- Physical handicaps such as motor difficulties (rare)

Patients may have one or more of these conditions in varying degrees of severity. In its most devastating form, TS renders the patient completely helpless and dependent.

Tuberous sclerosis was first identified in the late 1800's by Dr. Désiré-Magloire Bourneville, a French neurologist. Despite being one of the more common genetic disorders, TS remains poorly understood. The disorder is present at birth, but the clinical signs may be subtle and full symptoms may take considerable time to develop. As a result, TS frequently goes unrecognized or misdiagnosed for many years.

How common is tuberous sclerosis?

The true incidence of TS is unknown. Estimates of new cases of TS each year (the incidence rate) range from 1 in 10,000 to 1 in 30,000 births. However, the incidence could be greater, since many cases probably go unrecognized. The disorder affects both sexes and all races.

What causes tuberous sclerosis?

Tuberous sclerosis is caused by a defective gene that is usually inherited from a parent who has the

disorder. In some cases, however, the parent does not have TS but passes on a defective gene that has resulted from a spontaneous change or mutation. The cause of such a gene mutation is unknown.

The disease is inherited as an autosomal dominant disorder. This means that each child of an affected parent, or of a parent who is unaffected but carries the defective gene, has a 50 percent chance of developing symptoms.

What are the signs?

The signs of the disorder may vary according to the extent to which a particular body system is involved. Seizures, motor difficulties, developmental delay, mental retardation, hyperactivity, and behavioral abnormalities reflect brain involvement. Small benign tumors called angiofibromas sometimes form on the face. More commonly, white spots appear on the skin. These spots are of great importance in early diagnosis since they are present at birth in up to 90 percent of affected infants. The white spots are less pigmented than the adjacent skin, and can occur anywhere on the body.

What research is being done?

The National Institute of Neurological and Communicative Disorders and Stroke, a part of the National Institutes of Health, supports basic laboratory studies that may lead to a greater understanding of the causes of TS. Several scientists are searching for improved methods of treating the symptoms. Other investigators are trying to identify the location of the defective gene responsible for the disorder. Such a discovery would aid in diagnosis and treatment.

Clues about tuberous sclerosis may come from research on another genetic disease, neurofibromatosis. The two diseases—both of which cause tumors and skin spots—are believed to result from similar disruptions in the development of the central

nervous system. Some scientists are studying a naturally occurring fish model of neurofibromatosis in the hope of finding the cause of the disease. Other investigators are evaluating the effect of certain drugs on tumor growth. Advances achieved through such studies might benefit TS patients as well as those who have neurofibromatosis.

Is there any treatment?

Although there is no cure for TS, there are various forms of treatment for its symptoms. Recent studies have led to treatment advances in several areas. New anticonvulsant drugs are controlling seizures more effectively. Children with learning disabilities and speech and language difficulties are being helped through more sophisticated educational techniques. Teams of neurologists, orthopedists, physical therapists, occupational therapists, and other professionals are helping handicapped patients learn daily living skills and ways to improve mobility. With these and other advances, the outlook for tuberous sclerosis patients continues to improve.

Prognosis

The effect of tuberous sclerosis on a person's life varies according to the symptoms shown. Patients with the mild form of this disorder may live a normal and productive life. In the more severe forms of TS the disabilities may be serious, and premature death may occur as a result of infections, seizures, or tumors in vital organs.

How can I help research?

The National Institute of Neurological and Communicative Disorders and Stroke and the National Institute of Mental Health support a national human specimen bank at the Wadsworth Veterans Administration Hospital in Los Angeles. This bank supplies investigators around the world with tissue from patients with neurological and psychiatric diseases.

The bank needs tissue from tuberous sclerosis patients to enable scientists to study this disorder more intensely. Prospective donors should write to:

Dr. Wallace W. Tourtellotte, Director
Human Neurospecimen Bank
VA Wadsworth Medical Center
Building 212, Room 31
Los Angeles, CA 90073
Telephone: (213) 824-4307 (Call collect.)

Sources of information

Parents and patients who have TS can be kept informed of current research and medical advances by their physicians.

In addition, there are two nonprofit, tax-exempt organizations which provide information and support to families affected by the disorder:

American Tuberous Sclerosis Association
P.O. Box 44
Rockland, MA 02370
(617) 878-5528 or (toll free) 1-800-446-1211

National Tuberous Sclerosis Association
P.O. Box 612
Winfield, IL 60190
(312) 668-0787

For additional information concerning research supported by the National Institute of Neurological and Communicative Disorders and Stroke, contact:

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Prepared by
Office of Scientific and Health Reports
National Institute of Neurological and
Communicative Disorders and Stroke
Bethesda, Maryland 20892

NIH Publication No. 85-1846
August 1985

U.S. DEPARTMENT OF HEALTH
AND HUMAN SERVICES
Public Health Service
National Institutes of Health